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论著

Iα1基因hs1,2 区B等位基因与IgA肾病的易感性相关

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摘要

［摘要］ 目的：研究Iα1 hs1,2 VNTR多态性与我国IgA肾病的相关关系。 方法： 采集419例肾活检证实的IgA肾病患者及其一级亲属、条件相当的201例健康志愿者血样，提取基因组DNA。用PCR产物直接电泳法鉴定Iα1 hs1,2 VNTR基因型，采用以家庭为基础的传递/不平衡分析（TDT）和单倍型相对危险度（HRR），以及病例-对照研究分析Iα1 hs1,2 VNTR多态性与我国IgA肾病的相关关系。 结果： ① TDT分析结果显示Iα1 hs1,2 VNTR B等位基因从杂合子父母向患者传递的频率显著高于预期值(101 Trios, χ2=6.818, P＜0.01，扩展TDT分析也得到相同结果(164家庭, χ2=7.583, P＜0.01)。②与TDT结果一致，HRR分析同样显示Iα1 hs1,2 VNTR B等位基因的过度传递 (P＜0.05, χ2=4.122, HRR=1.180)， 而BB基因型具有更强的患病倾向 (P＜0.05, χ2=4.411, OR=1.538)。③病例-对照研究显示IgA肾病组B 等位基因频率显著高于正常对照组(χ2=6.968, P＜0.05)。 结论： Iα1 hs1,2 VNTR基因多态性与我国IgA肾病患者的易感性相关。

关键词 肾小球肾炎,IGA； 免疫球蛋白A； 增强子元件(遗传学)； 可变数量串联重复； 多态性现象(遗传学)； 传递不平衡分析

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B allele in Iα1 hs1,2 VNTR region is associated with IgA nephropathy

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Abstract

<FONT face=Verdana>AIM: To investigate the relationships between Iα1 hs1,2 VNTR polymorphism and IgA nephropathy. METHODS: Four hundred and nineteen patients with IgA nephropathy and their first-degree relatives were recruited. Two hundred and one sex and age-matched normal Chinese Han volunteers were also recruited as controls. After extracting genomic DNA, the VNTR genotypes of Iα1 hs1,2 region were determined by PCR and electrophoresis, and the results were analyzed by transmission disequilibrium test (TDT) and haplotype relative risk (HRR) in the families, and Chi-Square test in the case-control analysis. RESULTS: ① TDT analyses showed that B allele of the Iα1 hs1,2 VNTR region was significantly more transmitted from heterozygous parents to patients than expected (101 Trios, χ2=6.818, P<0.01), extended TDT produced the same results (164 families, χ2=7.583, P<0.01). ② Consistent with the TDT results, HRR also showed that B allele was over-transmitted to patients (P<0.05, χ2=4.122, HRR=1.180), and the BB genotype conferred a higher risk of developing the disease (P<0.05, χ2=4.411, OR=1.538). ③ The case-control study indicated that the B allele had a higher frequency in the IgA nephropathy group (χ2=6.968, P<0.05). CONCLUSION: B allele in Iα1 hs1,2 VNTR region is associated with susceptibility to IgA nephropathy.</FONT>

Key words Glomerulonephritis IGA； Immunoglobulin A Enhancer elements (genetics) Variable numbers of tandem repeats； Polymorphism (genetics) Transmission/Disequilibrium Test

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